

Parent Fact Sheet

Propionic Acidemia (PA)

Cause

Propionic Acidemia is one type of organic acid disorder. People with PA have problems breaking down and using certain amino acids from the foods they eat. PA occurs when an enzyme called “propionyl CoA carboxylase” is either missing or not working properly. When this enzyme is not working, substances called glycine and Propionic acid along with other harmful substances build up in the blood and cause problems.

If Not Treated

The symptoms can start as early as the first week of life. Symptoms include: extreme sleepiness or lack of energy, poor appetite, vomiting, low muscle tone, ketones in the urine, high blood ammonia levels, high blood levels of glycine, high levels of certain organic acids, low platelets, low white blood cells and increased levels of acidic substances in the blood called metabolic acidosis. If a metabolic crisis is not treated, a child with PA can develop: breathing problems, stroke, swelling of the brain, seizures and coma-sometimes leading to death.

A metabolic crisis can be triggered by eating large amounts of protein, illness or infection, going too long without food and stressful events such as surgery.

Treatment Options

Your doctor will work with a metabolic specialist and dietitian to care for your child. Lifelong treatment is usually needed.

Avoid going a long time without food. These babies need to eat more often to avoid low blood sugar. They should not go without eating for more than 4-6 hours. Some babies will need to eat more often than this. A low protein diet is often recommended. Your dietitian will help plan any diet changes.

Medical formula and foods contain the correct amount of protein and nutrients needed to for normal growth and development. Your metabolic doctor will tell you what type of formula is best and how much to use.

A medication that has been found to be beneficial is L-Carnitine. It is a safe and natural substance that helps the body create energy and rid the body of harmful wastes. Unless you are advised otherwise, use only L-Carnitine prescribed by your doctor. Some children may be prescribed Biotin supplements, which is a type of B vitamin.

If Treated

It is not known how effective treatment is in preventing problems. Children who need treatment are treated early may be able to live healthy lives with typical growth and development. Some children may continue to have seizures, mental retardation, involuntary movements and other health problems despite treatment.

For more information go to the following website: <http://www.newbornscreening.info>